



2018 Annual Report

A cure is out there.
Help us find it.



Our Impact this year

2018 Year In Review

The children and families depending on us to fulfill our mission are at the heart of every decision we make and every goal we set.

This year we made great progress in all areas of our mission. We awarded our largest single research grant to date, organized a successful International PCDH19 Professional and Family Conference, and sent 5 deserving families to the PCDH19 Conference for a life-changing experience. The PCDH19 Alliance and the progress we have made would not exist without our generous donors and fundraisers, and the hard work of our dedicated volunteers. Look what we've accomplished together this year.

Our Mission

Our mission is to improve the lives of children and families who are affected by PCDH19 Epilepsy. The Alliance focuses on raising and directing funds to scientific research with the goal of finding better, more effective treatments and, ultimately, a cure; providing information and support to affected families; and assisting the efforts of the medical community, so that no family suffers without a diagnosis and the most appropriate medical treatment.

PCDH19 Epilepsy

PCDH19 Epilepsy is a condition with a wide spectrum of severity in seizures, cognitive delays, and other symptoms, which are all caused by a mutation of the PCDH19 gene on the X chromosome. Males with the mutation, who will be largely unaffected, will pass the mutation on to 100% of their daughters and none of their sons. Women with the mutation have a 50% chance of passing it to their daughters and will pass it to 50% of their sons. Recently, scientists have discovered some unaffected females and are studying to learn what is protecting them from the disorder. Mosaic males are also affected, but so far very few males have been diagnosed.

BOARD OF DIRECTORS

JULIE WALTERS, PRESIDENT

KARIN WELLS-KILPATRICK, VICE-PRESIDENT

LESLIE HENKEL, SECRETARY

HEATHER FRYMAN, TREASURER

SUSAN LOFTUS TAYLOR

JENNIFER HOLLAND

SCIENTIFIC ADVISORY BOARD

DR. JOSEPH E. SULLIVAN, M.D.

DR. JACK PARENT, M.D.

PROFESSOR INGRID SCHEFFER

PROFESSOR JOZEF GECZ

Research Grants

We believe research brings us hope, and it is by far our largest program here at the Alliance. With the guidance of our esteemed International Scientific Advisory Board, we are funding cutting edge PCDH19 research every year. This year we funded our largest single grant to date. We received several scientifically strong proposals, and wish we could have funded them all. You can read about the project we funded below. We are looking forward to hearing Dr. Parent and Dr. Niu speak at our next conference.

Our goal is to fund research directly targeted toward understanding the expression of the PCDH19 gene and the function of the PCDH19 protein, finding therapeutic treatments, and a cure for PCDH19 Epilepsy.

We also hope that projects we fund will lead to additional research support from government or other funding agencies. We are pleased to be able to support many different types of projects, each critical for advancing all phases of PCDH19 research, from basic, to clinical, to treatment.



Modeling PCDH19-Related Epilepsy in human iPSC derived neurons and cerebral organoids

- Jack Parent, MD and Wei Niu, PhD

“PCDH19-Related Epilepsy (PRE) is caused by mutations in the PCDH19 gene located on the X-chromosome. How PCDH19 mutations result in epilepsy is poorly understood, as is the function of PCDH19 during human brain development. We use human pluripotent stem cells obtained by reprogramming patient skin cells or by CRISPR gene editing to generate brain cells that model PRE in cell culture. The studies described in this grant will take advantage of state-of-the-art stem cell approaches, including growing 3D “mini-brains” (cerebral organoids) in a dish, to determine the function of PCDH19 and how its mutations lead to seizure-like activity. Progress in our studies will help to identify PRE-related abnormalities in brain development and seizure mechanisms that should lead to novel therapies.”



Patient & Family Support

Patient Assistance

Besides the emotional and physical impact that PCDH19 has, the financial strain on a family living with PCDH19 Epilepsy can be devastating in itself. Travelling to our Biennial Research and Family conference, may not be an affordable option for them. We believe that bringing families together to hear the latest PCD19 research, interact with researchers, ask questions that are important to them, and bond with other families on the same journey is empowering and life changing. Many children meet others for the first time who are facing some of the same challenges. Because of our incredibly generous supporters, we were thrilled to provide travel and lodging for five PCDH19 families this year. We'd like to introduce you to them:



The Waddington Family (UK) — “Receiving confirmation that my family and I had been awarded a grant towards travel and accommodation to attend the pcdh19 conference 2018 was an amazing surprise to us and one we were so grateful for... It provided us with hope and excitement that advances have been made already and are continuing to be made.”



The Pendergraft Family (US) — “I am truly grateful for the opportunity to attend the Pcdh19 conference... It was motivating and inspiring... My favorite part was listening to the researchers from around the world talk about their cutting edge research.”

Patient & Family Support (cont'd)



The Lentz Family (US) — “I didn’t see any way of being able to go... We are so thankful for this opportunity to go to the conference. Finally, I have friends who truly understand and I no longer feel alone.”



The Lucchini Family (US) — “We all cried, laughed and shared our experience, strength and hope with each other... Being a single Mom and not working, I wouldn't have been able to attend the PCDH19 conference without the grant. “



The Otero-Green Family (US) — “Traveling to California seemed impossible... Thank you so much to the donors and everyone who made this trip possible . Our family is forever grateful for this amazing learning opportunity and the long lasting connections.”

Facebook Support Group



Our Facebook support group continues to grow rapidly. We added an average of 1 family per week in 2018. Our group continues to grow while remaining a safe, non-judgmental home where families can go to celebrate, commiserate, vent, ask questions, find all kinds of support, and find the latest information about PCDH19 Epilepsy and the PCDH19 Alliance. PCDH19 can be isolating, and this group is a lifeline and a home. Alliance volunteers administer the group, and must check in several times daily and also screen new member requests.

Education, Awareness, and Advocacy

Our Awareness and Education program continued to grow and make an impact on the medical community and patients waiting for a diagnosis.

This was our 6th year exhibiting at the American Epilepsy Society (AES) Conference, this year in New Orleans, where over 5000 medical professionals and researchers from around the world convened to exchange ideas and learn the latest information about epilepsy.

At AES we spend the bulk of our time educating medical professionals about the importance of a diagnosis and genetic testing, meeting with corporate partners, attending meetings, and reaching out to researchers. We do some of our most important work at this meeting each year, and this year was no exception.



A much loved part of our Patient Assistance and Support program has been our bear program, in which we send PCDH19 Alliance Bears to affected children and families. This year, thanks to an amazing PCDH19 Alliance volunteer mom and daughter duo, Cindy and Emily Beers, we were able to expand our Bear program to ensure each child newly diagnosed with PCDH19 has the opportunity to receive at no cost our sweet, cuddly and comforting symbol of hope - HOPE the bear. The total number of bears sent was 56, 38 domestically and 18 international shipments! We shipped



to 8 countries (including the US) and to 26 states in the US. International shipments went as far away as Australia and India! Thank you Cindy and Emily! Emily lives with PCDH19 and is finding meaning and spreading Hope to children.



Education, Awareness, and Advocacy (cont'd)

We were honored to be included with the Pasca Lab at Stanford in *Minds Wide Open*, an award winning documentary commissioned by the Chen Foundation. The Pasca Lab is a past recipient of a PCDH19 research grant. Professor Sergiu Pasca and his team's work focus specifically researching PCDH19 brain region-specific organoids to better understand PCDH19.



About *Minds Wide Open*

Husband and wife philanthropists, Tianqiao Chen and Chrissy Luo, commissioned Tim May, an award-winning producer who started his career at the BBC, to oversee production of the film with Merchant Cantos, a leading production company in the United Kingdom. The original soundtrack, composed by Sion Trefor, is brought to life by acclaimed singer-songwriter Charlotte Church. More information about the film can be found at www.MindsWideOpenFilm.com.

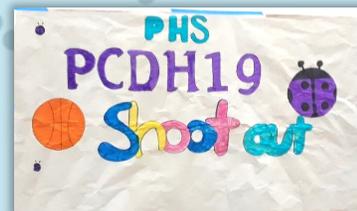
About the Tianqiao and Chrissy Chen Institute:

The Tianqiao and Chrissy Chen Institute (TCCI) was founded in 2016 by Tianqiao Chen and his wife Chrissy Luo, the founders of Shanda Group, who committed US \$1 billion to help advance fundamental brain research. With a US \$115 million donation, TCCI created the Tianqiao and Chrissy Chen Institute for Neuroscience at Caltech later that year. The couple has committed CNY 500 million to support TCCI's work in China and in 2017, they created a strategic partnership with the Zhou Liangfu Foundation and Huashan Hospital in Shanghai. In 2018, Shanghai Mental Health Center joined the partnership. Follow TCCI news at www.ChenInstitute.org, on LinkedIn, or via Twitter @ChenInstitute.

Our families and their friends in Canada, Poland, the U.S. and the U.K. continued to support the Alliance with creative fundraisers fueled by a desire to save the life of, ease the suffering of, and create a better future for their loved ones living with PCDH19. Our supporters organized basketball tournaments, PTO fundraisers, runs, walks, mountain climbing, online fundraising pages, speedway-race, chili-cook-offs, Facebook fundraisers, hockey games, bake sales, garage sales and the list goes on.



These fundraisers give families a voice, a way to raise awareness, and a way to contribute to science, to patient support and to fight for an effective treatment for their children. They are vital to making the research, the conferences and the support we offer possible. The time, effort and love that goes into these events. The courage shown by those willing to take action on behalf of the Alliance is inspiring and truly life changing.



PCDH19 Professional & Family Conference

Our 3rd Biennial PCDH19 Professional and Family Conference, was the highlight of the year for the PCDH19 Alliance Community. Researchers and families from 3 continents and several countries came together to present and discuss the latest PCDH19 research, share stories, and make friendships that will surely last a lifetime. This year we recorded the conference and have made the videos available on our website for everyone to view at their convenience.

A heartfelt thank you to Joseph Sullivan MD for volunteering his time each year to Chair this amazing and life-changing conference for the Alliance. Dr. Sullivan, who is also the Chair of our Scientific Advisory Board, volunteers a significant amount of his time organizing, planning, and hosting the conference and because of his dedication, the dedication of Tym Peters at UCSF, our Alliance Volunteers, and our sponsors, The PCDH19 Alliance has created a highly regarded research and family conference.



The Numbers

Raising and Investing Funds

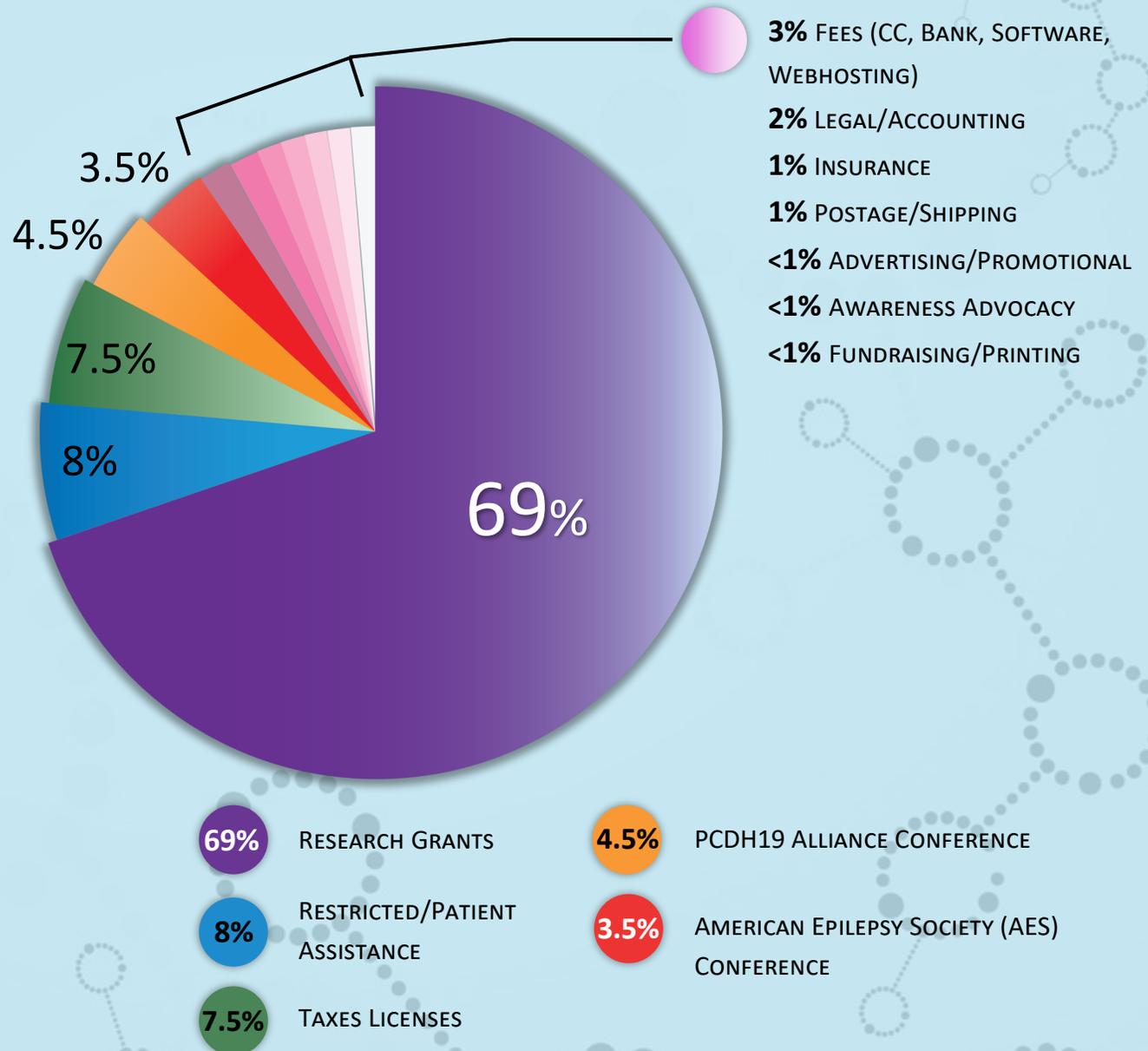
As an organization that serves and represents a rare disease, we are making a relatively large impact. Most of our donations continued to come from PCDH19 Families making personal donations and reaching out to family and friends to join our cause through various fundraisers.

With the guidance of our Scientific Advisory Board, we continued to fund the most promising PCDH19 research in 2018 and are excited to have the researchers update us all at our Professional and Family Conference in August 2020.

We continued to make a substantial impact by strategically leveraging funds and relying 100% on volunteers to travel, attend medical conferences, create awareness and educational materials, forge relationships with industry partners, and run our programs every day.

We are inspired and grateful that you have joined us to make a real impact for individuals and families living with PCDH19 Epilepsy. We take stewardship of your hard earned funds very seriously. Here are the numbers:

EXPENDITURES



Thank you

We are deeply grateful to all of our supporters and we were honored to recognize Lacey Smith, MS, CGC and Le Shepard with special awards.



Spirit of Progress Award

We recognized Lacey Smith with our Spirit of Progress Award for her dedication and commitment to creating and running the PCDH19 Registry, which is located at Boston Children's Hospital and UCSF. Lacey has gone above and beyond the call of duty on every occasion and help us find answers for our children and for adults living with PCDH19. Her commitment to our PCDH19 families is inspiring!



Spirit of Service Award

We recognized Le Shepard for her volunteer service to the PCDH19 Alliance over the years. Le has consistently stepped out of her comfort zone to assist the Alliance in our mission. She has hosted online fundraisers, lent her skills as a writer and a therapist to create relatable and well-crafted blog posts for our Alliance families, helps us raise awareness and runs our Alliance blog live from our biennial conference, so that those not in attendance can follow along or get the highlights at their convenience. Le has also volunteered at the American Epilepsy Society with us twice. Whenever we ask for help, Le's hand goes up.

Major Donors / Supporters

The Homewood Family

Cindy and Bobby Beers

Robert and Kay McManus

Robert and Francine Goldfarb

Kris and Tom Atkins

Pamela and Jack McManus

Karin and Bart Kilpatrick

Seema and James Adams

The Coultas Family

Julie Walters and Matt McManus

Jennifer and Tony Holland

Susan and Chuck Taylor

Kathy and Scott Tenchka

Charlene, Brookes and the Allan Family

April and James Haganey

The Dunn Family

Tom and Christina Lawson

Denis and Jennifer Austin

Robert and Susan Wells

The Pacifica High School Boys Basketball Team

Center Street PTO

Checking for Charity and the Dumont and Taylor Families

Family and Friends in Memory of Beverly Henkel

Family and Friends in Memory of Robin Dunn

Family and Friends in Memory of Suzanne Haganey